



## tetra-amelia syndrome

Tetra-amelia syndrome is a very rare disorder characterized by the absence of all four limbs. ("Tetra" is the Greek word for "four," and "amelia" refers to the failure of an arm or leg to develop before birth.) This syndrome can also cause severe malformations of other parts of the body, including the face and head, heart, nervous system, skeleton, and genitalia. The lungs are underdeveloped in many cases, which makes breathing difficult or impossible. Because children with tetra-amelia syndrome have such serious medical problems, most are stillborn or die shortly after birth.

### Frequency

Tetra-amelia syndrome has been reported in only a few families worldwide.

### Genetic Changes

Researchers have found a mutation in the *WNT3* gene in people with tetra-amelia syndrome from one large family. This gene is part of a family of WNT genes that play critical roles in development before birth. The protein produced from the *WNT3* gene is involved in the formation of the limbs and other body systems during embryonic development. Mutations in the *WNT3* gene prevent cells from producing functional WNT3 protein, which disrupts normal limb formation and leads to the other serious birth defects associated with tetra-amelia syndrome.

In other affected families, the cause of tetra-amelia syndrome has not been determined. Researchers believe that unidentified mutations in *WNT3* or other genes involved in limb development are probably responsible for the disorder in these cases.

### Inheritance Pattern

In most of the families reported so far, tetra-amelia syndrome appears to have an autosomal recessive pattern of inheritance. Autosomal recessive inheritance means both copies of the gene in each cell have mutations. The parents of an individual with tetra-amelia syndrome each carry one copy of the mutated gene, but do not show signs and symptoms of the condition.

### Other Names for This Condition

- Tetra-amelia
- Tetra-amelia, autosomal recessive

## **Diagnosis & Management**

### Genetic Testing

- Genetic Testing Registry: Tetraamelia, autosomal recessive  
<https://www.ncbi.nlm.nih.gov/gtr/conditions/C2749279/>

### Other Diagnosis and Management Resources

- GeneReview: Tetra-Amelia Syndrome  
<https://www.ncbi.nlm.nih.gov/books/NBK1276>

### General Information from MedlinePlus

- Diagnostic Tests  
<https://medlineplus.gov/diagnostictests.html>
- Drug Therapy  
<https://medlineplus.gov/drugtherapy.html>
- Genetic Counseling  
<https://medlineplus.gov/geneticcounseling.html>
- Palliative Care  
<https://medlineplus.gov/palliativecare.html>
- Surgery and Rehabilitation  
<https://medlineplus.gov/surgeryandrehabilitation.html>

## **Additional Information & Resources**

### MedlinePlus

- Health Topic: Bone Diseases  
<https://medlineplus.gov/bonediseases.html>

### Genetic and Rare Diseases Information Center

- Tetra-amelia syndrome  
<https://rarediseases.info.nih.gov/diseases/5148/tetra-amelia-syndrome>

### Educational Resources

- Disease InfoSearch: Tetraamelia autosomal recessive  
<http://www.diseaseinfosearch.org/Tetraamelia+autosomal+recessive/7034>
- Lucile Packard Children's Hospital: Congenital Limb Defects  
<http://www.stanfordchildrens.org/en/topic/default?id=congenital-limb-defects-90-P02752>

- MalaCards: tetra-amelia syndrome  
[http://www.malacards.org/card/tetra\\_amelia\\_syndrome](http://www.malacards.org/card/tetra_amelia_syndrome)
- Orphanet: Tetraamelia-multiple malformations syndrome  
[http://www.orpha.net/consor/cgi-bin/OC\\_Exp.php?Lng=EN&Expert=3301](http://www.orpha.net/consor/cgi-bin/OC_Exp.php?Lng=EN&Expert=3301)

#### Patient Support and Advocacy Resources

- Resource list from the University of Kansas Medical Center: Limb Anomalies  
<http://www.kumc.edu/gec/support/limb.html>
- The Compassionate Friends  
<https://www.compassionatefriends.org/>

#### GeneReviews

- Tetra-Amelia Syndrome  
<https://www.ncbi.nlm.nih.gov/books/NBK1276>

#### Scientific Articles on PubMed

- PubMed  
<https://www.ncbi.nlm.nih.gov/pubmed?term=%28%28tetra-amelia%5BTIAB%5D%29+OR+%28tetraamelia%5BTIAB%5D%29%29+AND+english%5Bla%5D+AND+human%5Bmh%5D>

#### OMIM

- TETRAAMELIA SYNDROME, AUTOSOMAL RECESSIVE  
<http://omim.org/entry/273395>

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